17th INTERNATIONAL HL7 INTEROPERABILITY CONFERENCE IHIC 2017

Hochschule Niederrhein
University of Applied Sciences
Gesundheitswesen
Faculty of Health Care

Standardizing the Transmission of Genomic-Related Medical Data for the Optimization of Diagnostics and Research in the Field of Oncological Diseases

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17th International HL7 Interoperability Conference 2017
Athens | Greece | Oct 22-24 2017

Background

- In the context of cancer treatment, there is a recent movement from “intervention” to “prevention”
- Advances in molecular biology have brought new information about alternative therapy options
- Molecular-genetic diagnostics of tumor cells provide accurate information about possible entry points for efficient therapy options
- The new methods of genome sequencing are summarized by the concepts “Next Generation Sequencing“(NGS) and “Precision Medicine“
- Currently, there is no consistent analytic and data transmission structure within genomic diagnostics in oncological diseases
- Within the framework of the project GENeALYSE, a standardized and interoperable specification for associated use cases shall be developed

GENeALYSE project partners

Niederrhein University of Applied Sciences Krefeld

Competence Center eHealth

project lead and admin, expertise in standardization

University Hospital Dusseldorf

Center for familial breast- and ovarian cancer therapy (ZFBEK-D)

Genomic diagnostics for the detection of a predisposition in the area of gynecological tumor diseases for the derivation of preventive measures

University Hospial Cologne, Department of Pathology

Genomic diagnostics on solid tumors for the best possible attack points of chemotherapeutic agents

GENeALYSE project goals

- GENeALYSE is intended to optimize the coordination between the diagnostic genome laboratory and the clinical therapy decision in order to increase the safety and success of the cancer treatment

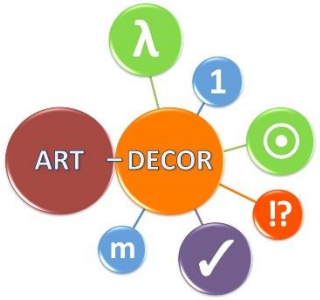
- Transmission of biomarker- and gene-analysis data to optimize the research base for tumor diseases

- Creation of an HL7 CDA R2 implementation guide for the optimization of the cooperation between diagnostics and therapy

GENeALYSE milestones

- Depiction of existing workflows in genome analysis (BPMN Diagrams)
- Capture of desiderata with regard to the medical experts (physicians and scientists)
- Development of a unique medical report structure as a basis for the specification
- Semantic annotation of datasets using SNOMED CT, LOINC, HGSV etc.
- HL7 CDA R2 Implementation Guide

Tooling with ART-DECOR®



- Software tool to enable cooperative work between healthcare professionals, terminologists and implementers
- Open source tool for the documentation and specification of CDA templates, value sets, scenarios and data sets
- Provides semantic annotations / Integrated terminology browsers
- Currently used in many European projects

www.art-decor.org

Preliminary Work

HL7 Implementation Guide for CDA® Release 2: Genetic Testing Report (GTR), DSTU Release 1

Purpose of the IG:

Specification of a document standard for the transmission of genetic test reports between laboratories and requesting environment (e.g. hospital)

Machine-processable analysis of results through unamgigous semantics

Enabling Decision Support and providing an improved research basis

Genetic Testing Report

Structure

- Summary Section

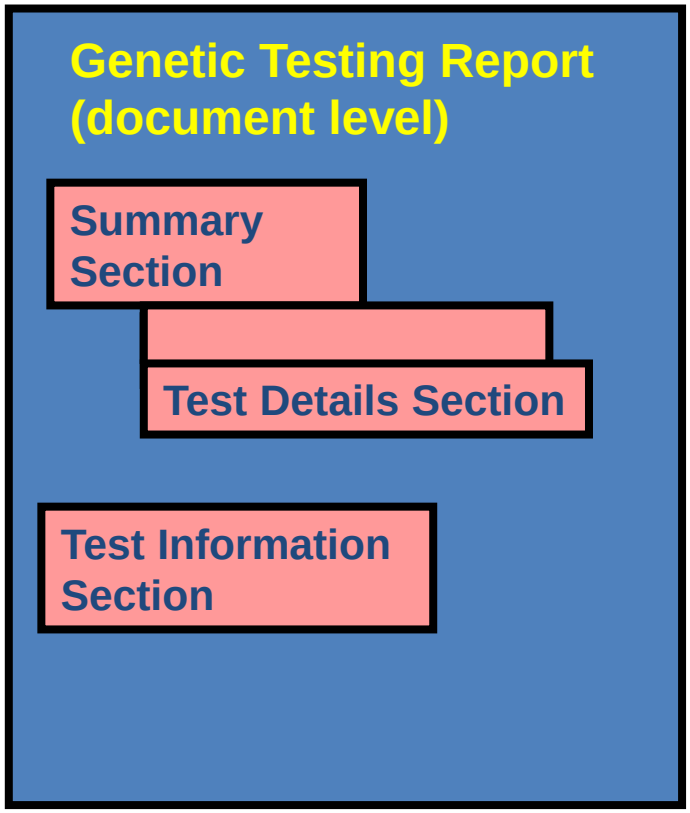
  Indication, Listing the performed tests, comprehensive interpretation, specimen

- Test Details Section

  Detailed description of performed tests

- Test Information Section

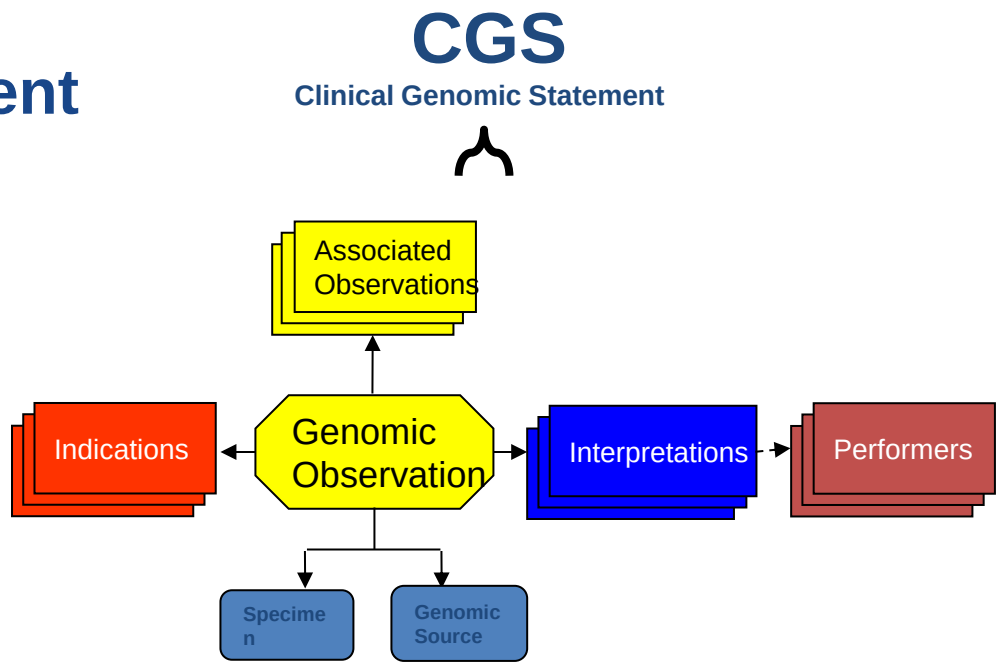
  Background information for performed tests, methods and references



Level 3 Entries

Clinical Genomic Statement (CGS) template

- Main template to provide structured data
- Unamgigous structure and semantics



Terminologies and Value Sets

LOINC

- Sections, Tests und Value Sets for interpretation of tests and results

SNOMED CT

- Additional information: Clinical findings, disorders, specimen

Human Genome Organization Nomenclature (HGNC) Committee

- DNA Gene Identifier – full specification of the gene

Human Genome Variation Society (HGVS) Nomenclature

- Description of sequence-variations

NCBI Database of Single Nucleotide Polymorphism (dbSNP)

- Sequence Variation Identifier (changes of nucleotids)

Synergies and related research

Campbell WS, Campbell JR: Binding SNOMED CT and Genomic Data for Cancer Care: An Implementation Story.

SNOMED CT Expo 2017 Bratislava

SCT obervable model to represent genomic data as collected by molecular pathologists using Next Generation Sequencing in the electronic health record and tissue biorepositories (University of Nebraska Medical Center)

- SNOMED CT concepts are deployed in the clinical information systems beginning in the laboratory and molecular pathology information systems and bound to molecular observations

Aso S et al. (2013): Analyzing SNOMED CT and HL7 terminology binding for semantic interoperability on post-genomic clinical trials. *Stud Health Technol Inform. 2013;192:980*

EU funded INTEGRATE project, to provide an infrastructure to share knowledge and data in post-genomic breast cancer clinical

Questions?
Thank you for your attention !

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